

Resource Summary Report

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HmtDB - Human Mitochondrial DataBase

RRID:SCR_007713

Type: Tool

Proper Citation

HmtDB - Human Mitochondrial DataBase (RRID:SCR_007713)

Resource Information

URL: <http://www.hmtdb.uniba.it>

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Description: A human mitochondrial resource aimed at supporting population genetics and mitochondrial disease studies. It consists of a database of Human Mitochondrial Genomes annotated with population and variability data, the latter estimated through the application of a new approach based on site-specific nucleotidic and aminoacidic variability calculation (SiteVar and MitVarProt programs). The goals of HmtDB are: to collect and integrate the publicly available human mitochondrial genomes data; to produce and provide the scientific community with site-specific nucleotidic and aminoacidic variability data estimated on all the collected human mitochondrial genome sequences; to allow any researcher to analyse his own human mitochondrial sequences (both complete and partial mitochondrial genomes) in order to automatically detect the nucleotidic variants compared to the revised Cambridge Reference Sequence (rCRS) and to predict their haplogroup paternity. HmtDBs first release contains 1255 human mitochondrial genomes derived from public databases (GenBank and MitoKor). The genomes have been stored and analysed as a whole dataset and grouped in continent-specific subsets (AF: Africa, AM: America, AS: Asia, EU: Europe, OC: Oceania). The multialignment and site-variability analysis tools included in HmtDB are clustered in two Work Flows: the Variability Generation Work Flow (VGWF) and the Classification Work Flow (CWF), which are applied both to human mitochondrial genomes stored in the database and to newly sequenced genomes submitted by the user, respectively., THIS RESOURCE IS NO LONGER IN SERVICE. Documented on September 16,2025.

Synonyms: HmtDB

Resource Type: data or information resource, database

Keywords: mitochondrial disease, mitochondrial dna, mtdna

Availability: THIS RESOURCE IS NO LONGER IN SERVICE

Resource Name: HmtDB - Human Mitochondrial DataBase

Resource ID: SCR_007713

Alternate IDs: nif-0000-02970

Record Creation Time: 20220129T080243+0000

Record Last Update: 20260815T182857+0000

Ratings and Alerts

No rating or validation information has been found for HmtDB - Human Mitochondrial DataBase.

No alerts have been found for HmtDB - Human Mitochondrial DataBase.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 34 mentions in open access literature.

Listed below are recent publications. The full list is available at [ASWG](#) .

Neueder A, et al. (2024) Huntington's disease affects mitochondrial network dynamics predisposing to pathogenic mitochondrial DNA mutations. *Brain : a journal of neurology*, 147(6), 2009.

Lin Y, et al. (2024) HiFi long-read amplicon sequencing for full-spectrum variants of human mtDNA. *BMC genomics*, 25(1), 538.

Wei R, et al. (2023) The mitochondrial genome as a modifier of autism versus cancer phenotypes in PTEN hamartoma tumor syndrome. *HGG advances*, 4(3), 100199.

Dunn PJ, et al. (2022) Investigation of Mitochondrial Related Variants in a Cerebral Small Vessel Disease Cohort. *Molecular neurobiology*, 59(9), 5366.

Zhang X, et al. (2022) Association of mitochondrial variants and haplogroups identified by whole exome sequencing with Alzheimer's disease. *Alzheimer's & dementia : the journal of the Alzheimer's Association*, 18(2), 294.

Gowri P, et al. (2022) Mutation profile of neurodegenerative mitochondriopathy - LHON in Southern India. *Gene*, 819, 146202.

Miglietta S, et al. (2022) MicroRNA and Metabolic Profiling of a Primary Ovarian Neuroendocrine Carcinoma Pulmonary-Type Reveals a High Degree of Similarity with Small Cell Lung Cancer. *Non-coding RNA*, 8(5).

- Crysap B, et al. (2021) Graph Algorithms for Mixture Interpretation. *Genes*, 12(2).
- Smart U, et al. (2021) A Continuous Statistical Phasing Framework for the Analysis of Forensic Mitochondrial DNA Mixtures. *Genes*, 12(2).
- Diroma MA, et al. (2021) New Insights Into Mitochondrial DNA Reconstruction and Variant Detection in Ancient Samples. *Frontiers in genetics*, 12, 619950.
- Zaidieh T, et al. (2021) Mitochondrial DNA abnormalities provide mechanistic insight and predict reactive oxygen species-stimulating drug efficacy. *BMC cancer*, 21(1), 427.
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- Hathazi D, et al. (2020) Metabolic shift underlies recovery in reversible infantile respiratory chain deficiency. *The EMBO journal*, 39(23), e105364.
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- Labory J, et al. (2020) Multi-Omics Approaches to Improve Mitochondrial Disease Diagnosis: Challenges, Advances, and Perspectives. *Frontiers in molecular biosciences*, 7, 590842.
- N??meth K, et al. (2019) Next-generation sequencing identifies novel mitochondrial variants in pituitary adenomas. *Journal of endocrinological investigation*, 42(8), 931.
- Picca A, et al. (2019) Mitochondrial-Derived Vesicles as Candidate Biomarkers in Parkinson's Disease: Rationale, Design and Methods of the EXosomes in PArkiNson Disease (EXPAND) Study. *International journal of molecular sciences*, 20(10).
- Guerra F, et al. (2019) Synergistic Effect of Mitochondrial and Lysosomal Dysfunction in Parkinson's Disease. *Cells*, 8(5).
- You C, et al. (2019) Mitochondrial DNA analyses found five novel mutations in idiopathic epilepsy patients. *Mitochondrial DNA. Part B, Resources*, 4(2), 2387.
- Starikovskaya E, et al. (2019) Mitochondrial DNA Variation of Leber's Hereditary Optic Neuropathy in Western Siberia. *Cells*, 8(12).